



CATSPER1 gene

cation channel sperm associated 1

Normal Function

The *CATSPER1* gene provides instructions for producing a protein that is found in the tail (flagellum) of sperm cells. The CATSPER1 protein plays a role in sperm cell movement (motility) and is required for sperm cells to push through the outside membrane of the egg cell during fertilization. The CATSPER1 protein is embedded in the membrane of sperm cells and is necessary for positively charged calcium atoms (calcium cations) to enter the cell. Calcium cations are required for a type of sperm motility called hyperactivation. Hyperactivation is characterized by vigorous movements of the sperm tail, which are necessary for the sperm to push through the membrane of the egg cell during fertilization.

Health Conditions Related to Genetic Changes

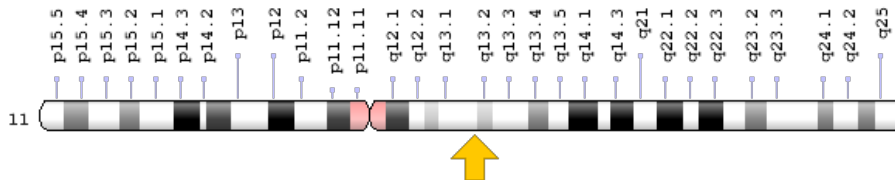
CATSPER1-related nonsyndromic male infertility

At least two mutations in the *CATSPER1* gene have been found to cause *CATSPER1*-related nonsyndromic male infertility. These mutations are thought to lead to the production of a CATSPER1 protein that may be altered, nonfunctional, or quickly broken down (degraded) by the cell. A lack of functional CATSPER1 protein impairs calcium entry into the sperm cell, which decreases motility and prevents hyperactivation. Lack of hyperactivation results in sperm that are unable to push through the membrane of the egg cell and achieve fertilization. These sperm abnormalities are the cause of infertility in affected males. Male infertility is the only symptom of *CATSPER1*-related nonsyndromic male infertility.

Chromosomal Location

Cytogenetic Location: 11q13.1, which is the long (q) arm of chromosome 11 at position 13.1

Molecular Location: base pairs 66,016,752 to 66,026,518 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cation channel sperm-associated protein 1
- cation channel, sperm associated 1
- CATSPER
- CTSR1_HUMAN
- sperm-associated cation channel 1
- sperm ion channel

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Action at a Distance: Mammalian Gametes
<https://www.ncbi.nlm.nih.gov/books/NBK10010/?rendertype=box&id=A1381>

GeneReviews

- CATSPER-Related Male Infertility
<https://www.ncbi.nlm.nih.gov/books/NBK22925>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CATSPER1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- CATION CHANNEL, SPERM-ASSOCIATED, 1
<http://omim.org/entry/606389>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CATSPER1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CATSPER1%5Bgene%5D>
- HGNC Gene Family: Cation channels sperm associated
<http://www.genenames.org/cgi-bin/genefamilies/set/186>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17116
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/117144>
- UniProt
<http://www.uniprot.org/uniprot/Q8NEC5>

Sources for This Summary

- Avenarius MR, Hildebrand MS, Zhang Y, Meyer NC, Smith LL, Kahrizi K, Najmabadi H, Smith RJ. Human male infertility caused by mutations in the CATSPER1 channel protein. *Am J Hum Genet.* 2009 Apr;84(4):505-10. doi: 10.1016/j.ajhg.2009.03.004. Epub 2009 Apr 2.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19344877>
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<http://omim.org/entry/606389>
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